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Leukaemia Section
Short Communication

t(4;12)(q21;p13)

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Abstract

Review on t(4;12)(q21;p13) in lymphoid malignancies

Keywords
Chromosome 4; Chromosome 12; Acute lymphoblastic leukemia; Primary mediastinal large B-cell lymphoma; Adult T-cell leukemia/lymphoma; Acute megakayoblastic leukemia.

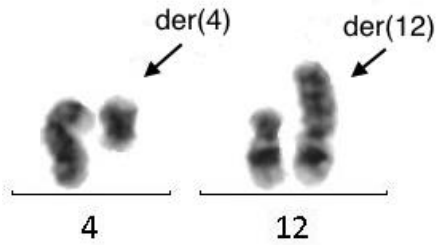

Figure 1. Partial karyotype with t(4;12)(q21;p13) in a female patient with B-ALL (table 1, #8).

Clinics and pathology

Disease

Translocation t(4;12)(q21;p13) occurs predominantly in B-cell lymphoid disorders, including ALL, NHL, rarely with T-ALL and AML.

Acute lymphoblastic leukemia (ALL): was diagnosed in 5 patients (Groupe Franais de Cytogntique Hmatologique, 1993; Behm et al., 1996; Elghezal et al., 2001; Gindina T., own case, table 1,#8).

Primary mediastinal large-B-cell lymphoma was found in 1 patient (Palanisamy et al, 2002)

Adult T-cell Leukemia: 1 patient (Sadamori et al., 1991).

Non-Hodgkins lymphoma: 1 patient (Schouten et al., 1990).

Acute myeloid leukemia: 1 patient with AML-M7 (Ohyashiki et al., 1984).

Phenotype/cell stem origin

Two patients had B-cell Early B ALL (CD10+) and Pre-B ALL (CIg +) (Groupe Franais de Cytogntique Hmatologique, 1993).

Epidemiology

The translocation t(4;12)(q21;p13) was found in adults and children as well as equally among male and female patients.

Genetics

Genes implicated in this translocation remain unknown.

Pts	Disease	Gender/Age	Karyotype
1	NHL	F/?	49,XX, +3,+18,t(3;?)(q29;?), **t(4;12)(q21;p13)**, del(6)(q23),t(7;?)(q2l;?),t(9;?)(p23;?),t(14;18)(q32;q21),t(17;?)(p13;?),+mar
2	ATL	M/?	48,XY,+del(l)(p32),+3,**t(4;12)(q21;p13)**
3	PMBL	F/?	50,XX, dup(2)(q21q31),+3,**t(4;12)(q21;p13)**, del(6)(q23),del(7)(q21),add(9)(p23),add(13)(p13),t(14;18)(q32;q21),+18, +add(20)(p13),+21
4	B-ALL	F/?	47,XX,**t(4;12)(q13;p12)**,del(15)(q14q25),+21,der(21) t(12;21)[3]/ 48,idem,+13[1]/49,idem,+X[1]
5	B-ALL Remission duration 30+	F/6	47,XX,t(1;12)(p22;p13),**t(4;12)(q21;p13)**,+10, del(11)(q23)/45,X,-X,t(4;12)
6	B-ALL	M/3	47,XY,del(1q),del(2q),der(3q), **t(4;12)(q13;p12)**, del(6)(q21q25), + mar
7	B-ALL	M/5	46, XY, der(1)t(1;?)(p36;?), **t(4;12)(q13;p12)**
8	B-ALL	F/3	48,XX,**t(4;12)(q21;p13)**,+10,+21
9	AML-M7	M/59	46,XY,**t(4;12)(q21;p13)**

Table 1. Reported cases with t(4;12)(q21;p13).
NHL: non-Hodgkin's lymphoma; ATL: Adult T-cell leukemia; PMBL: Primary mediastinal large-B-cell lymphoma; B-ALL: B-Acute lymphoblastic leukemia; AML-M7: Acute megakaryoblastic leukemia
1. Schouten et al., 1990: 2. Sadamori et al., 1991; 3. Palanisamy et al., 2002; 4. Elghezal et al., 2001; 5. Behm et al., 1996; 6,7. Groupe Franais de Cytogntique Hmatologique, 1993; 8. Gindina, personal observation; 9. Ohyashiki et al., 1984.

Cytogenetics

Additional anomalies

Additional chromosome abnormalities were observed in all these patients with lymphoid malignancies. Translocation t(4;12) was associated with such anomaly as translocation t(14;18)(q32;q21) in 2 cases (Schouten et al., 1990; Palanisamy et al., 2002) and translocation t(12;21)(p13;q22) ETV6/ RUNX1 in 1 patient (Elghezal et al., 2001).

Trisomies of chromosomes 3 and 21 were present in 3 patients each (Schouten et al., 1990, Sadamori et al., 1991; Palanisamy et al., 2002; and Palanisamy et al., 2002; Elghezal et al., 2001; Gindina T. own case #8, respectively), trisomies of chromosomes 10 and 18 were in 2 cases each (Behm et al., 1996; Gindina T. own case #8, respectively and Schouten et al., 1990; Palanisamy et al., 2002, respectively). Deletion of 6q was demonstrated in 3 patients (Schouten et al., 1990; Palanisamy et al., 2002; Groupe Franais de Cytogntique Hmatologique, 1993). In most cases, t(4;12)(q21;p13) was as a part of a complex karyotype. As a single anomaly, t(4;12)(q21;p13) was described only in one case of AML (Ohyashiki et al., 1984).

Result of the chromosomal anomaly

Fusion protein

Oncogenesis

Most likely, the translocation t(4;12)(q21;p13) is a secondary genetic event in oncogenesis.

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